Disease Name — Medium-chain acyl-CoA dehydrogenase deficiency

Alternate name(s)	None
Acronym	MCADD
Disease Classification	Fatty Acid Oxidation Disorder
Variants	N/A
Variant name	N/A
Symptom onset	Typically 6-24 months but ranges from neonatal to adult
Symptoms	Recurrent episodes of hypoglycemia, vomiting, coma, sudden death and possible seizures. Hepatomegaly usually present.
Natural history without treatment	Metabolic episodes can cause developmental and physical delays, neurologic impairment and sudden death.
Natural history with treatment	Normal intellect and physical functioning expected.
Treatment	Dietary: avoid fasting, low-fat diet (<30% of dietary fat), carnitine supplementation, cornstarch supplementation.
Emergency Medical Treatment	See sheet from American College of Medical Genetics (attached) or for more information, go to website: http://www.acmg.net/StaticContent/ACT/C8_C6_C10.pdf
Physical phenotype	None
Inheritance	Autosomal recessive
General population incidence	1/15,000
Ethnic differences	Yes
Population	Incidence higher in Northern Europeans and U.S Caucasians.
Ethnic incidence	Approximately 1/70 carrier rate
Enzyme location	Liver, heart, muscle and fibroblasts
Enzyme Function	Mitochondrial beta-oxidation of fat stores
Missing Enzyme	Medium-chain acyl-CoA dehydrogenase
Metabolite changes	Increased medium chain fatty acids, increased glycine/carnitine esters, increased dicarboxylic acids.
Prenatal testing	DNA and enzymatic testing
MS/MS Profile	Elevated C10:1, C8, C6
OMIM Link	http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=201450
Genetests Link	www.genetests.org
Support Group	FOD Family Support Group http://www.fodsupport.org Organic Acidemia Association Http://www.oaanews.org Save Babies through Screening Foundation http://www.savebabies.org Genetic Alliance http://www.geneticalliance.org

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